GALAXY REGISTRY
GENERATING ADVANCEMENTS IN LONGITUDINAL ANALYSIS IN X&Y VARIATIONS

AXYS 2025

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INTRODUCTION

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1876

Children’s Hospital Colorado
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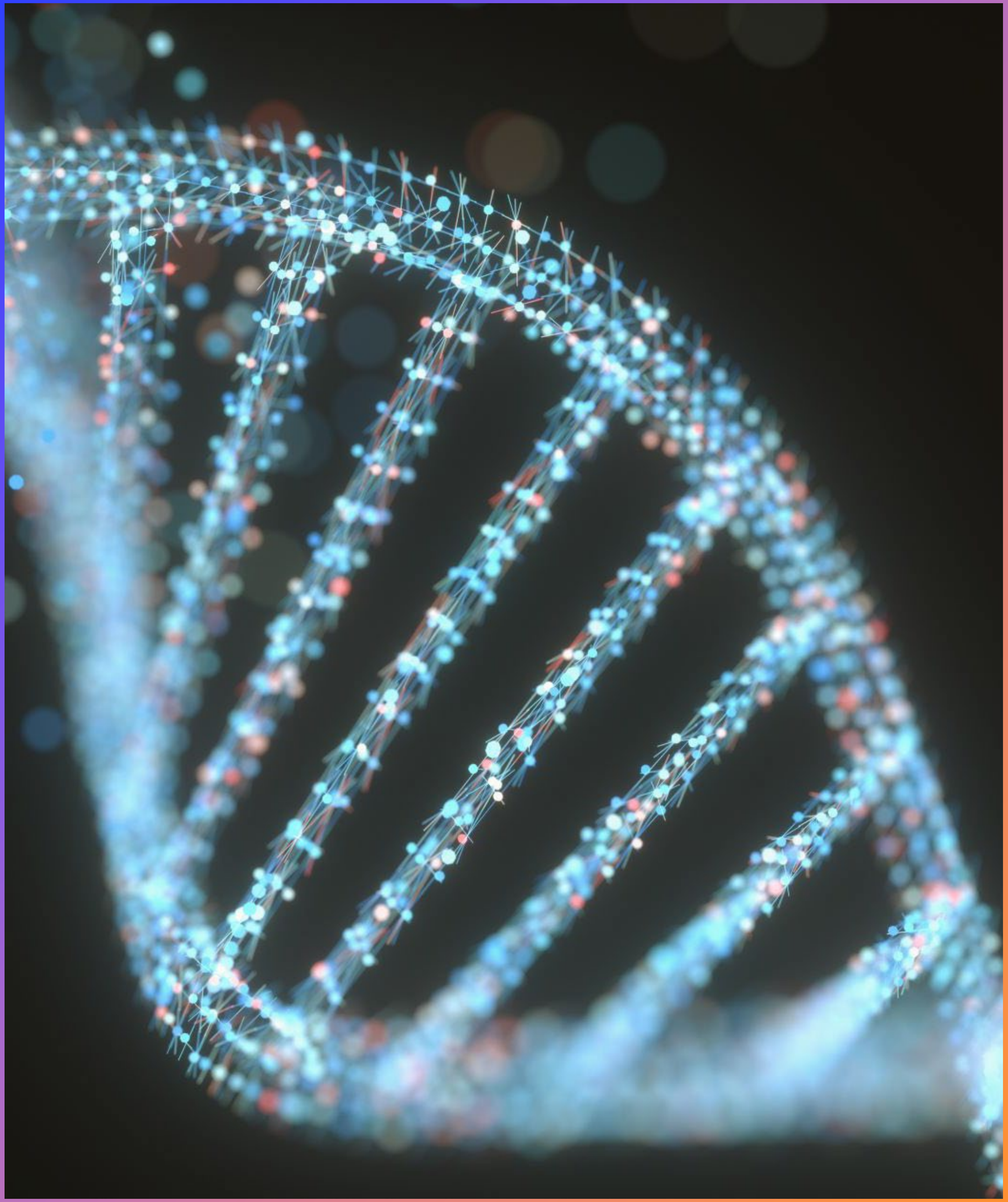
eXtraordinarY Kids

XXYY PROJECT

axys™



AGENDA

- What is GALAXY?
- Why is GALAXY important?
- How are we doing so far?
- Where are we going?
- Who will engage next?

WHAT IS GALAXY?

What is GALAXY?

Multicenter clinical data registry and biorepository for individuals with X&Y variations to…

- quantify baseline clinical outcomes
- determine best clinical practices
- support future research and quality improvement efforts

…that will ultimately improve outcomes for individuals with X&Y variations

What is GALAXY?

Joint Community Venture

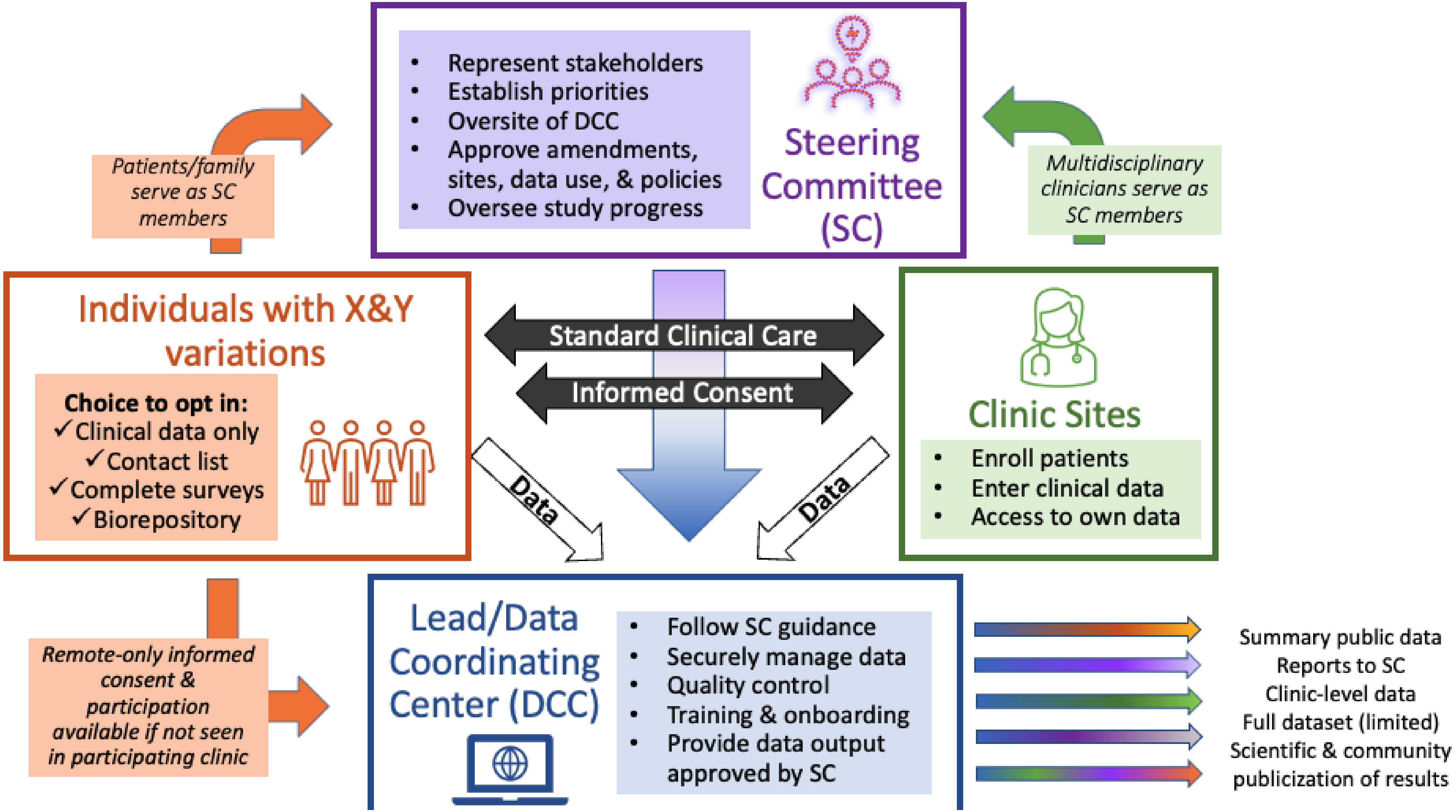
- Individuals with X&Y variations
- Parents & Partners
- Advocacy organizations
- Clinicians
- Researchers

Data Collection & Storage

- Health records
- Contact information
- Survey responses
- Blood specimens

Study Meaningful Things

- Ask the right questions the right way
- Prioritize resources to get results
- Ongoing dialog between community members



Steering Committee (SC)

- Represent stakeholders
- Establish priorities
- Oversite of DCC
- Approve amendments, sites, data use, & policies
- Oversee study progress

Patients/family serve as SC members

Multidisciplinary clinicians serve as SC members

Individuals with X&Y variations

Choice to opt in:
✓Clinical data only
✓Contact list
✓Complete surveys
✓Biorepository

Standard Clinical Care

Informed Consent

Data

Data

Clinic Sites

- Enroll patients
- Enter clinical data
- Access to own data

Remote-only informed consent & participation available if not seen in participating clinic

Lead/Data Coordinating Center (DCC)

- Follow SC guidance
- Securely manage data
- Quality control
- Training & onboarding
- Provide data output approved by SC

Summary public data
Reports to SC
Clinic-level data
Full dataset (limited)
Scientific & community publicization of results

Balance

Focus on specific aims
Targeted data collection
Limited time/scope
Clear products

Focus on infrastructure
Broad data collection
Longitudinal emphasis
Resource for future research

WHY IS GALAXY IMPORTANT?

We need to catch X&Y up!

X&Y variations are under-recognized, underdiagnosed, and understudied → we need to change this!

Individually, still “rare disease”, and there is power in numbers.

Organized data collection and respository

“Clinical trial readiness” – to successfully do intervention studies

HOW ARE WE DOING SO FAR?

Timeline

Prior to starting GALAXY: INSIGHTS as a model (150K funding)

Seed Funding from AXYS, XXYY Project, & Living with XXY

April 28, 2022
Regulatory approval

June 16, 2022
First participant at Children's Hospital Colorado enrolled

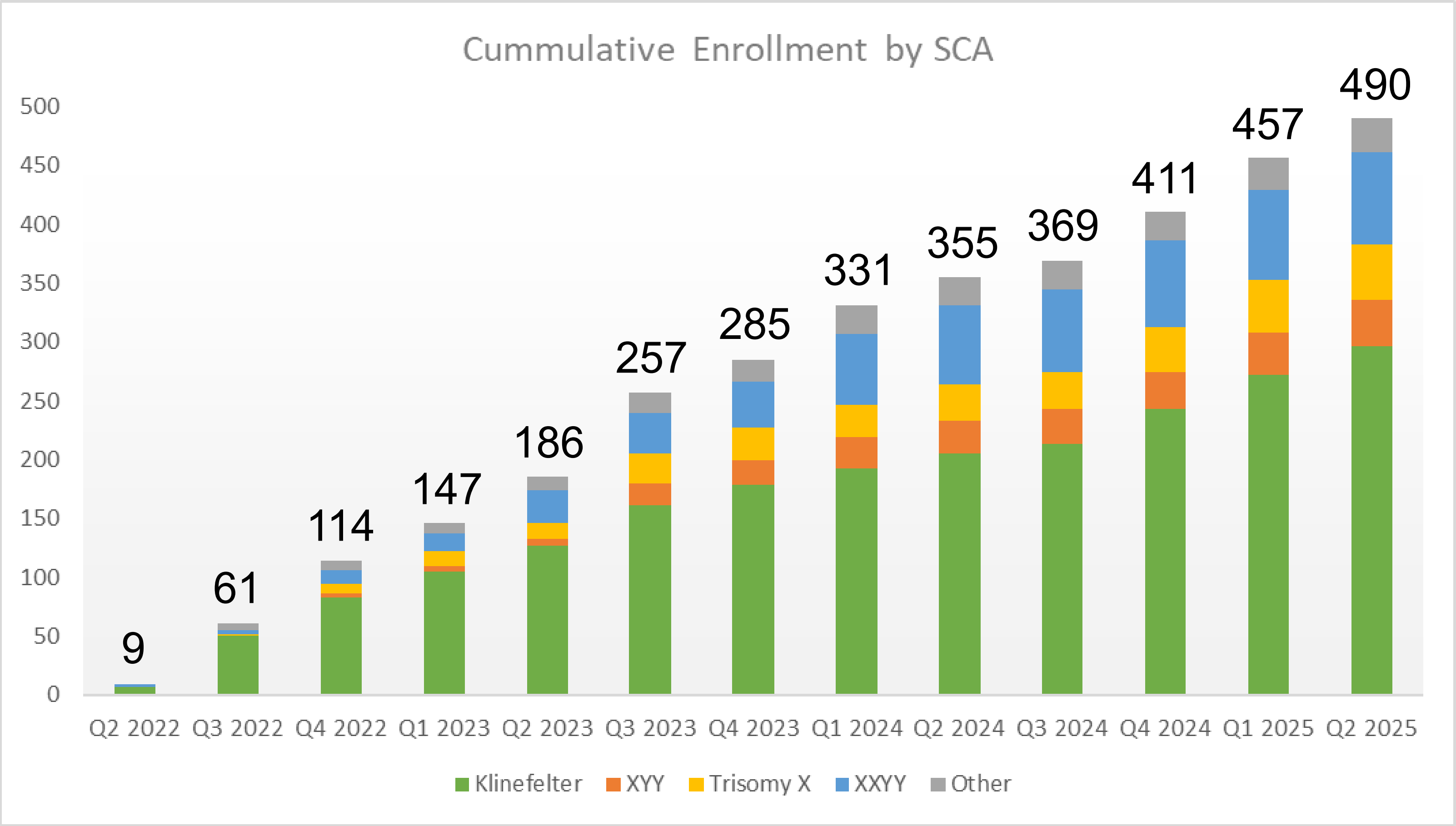
August 15, 2022
First participant outside of CHCO enrolled

October 7, 2022
First Steering Committee Meeting

May 16, 2024
Participants can enroll at Nemours Delaware and Lurie's Chicago

As of July 18, 2025
510 people have consented

Recruitment and Enrollment



XXY = 285

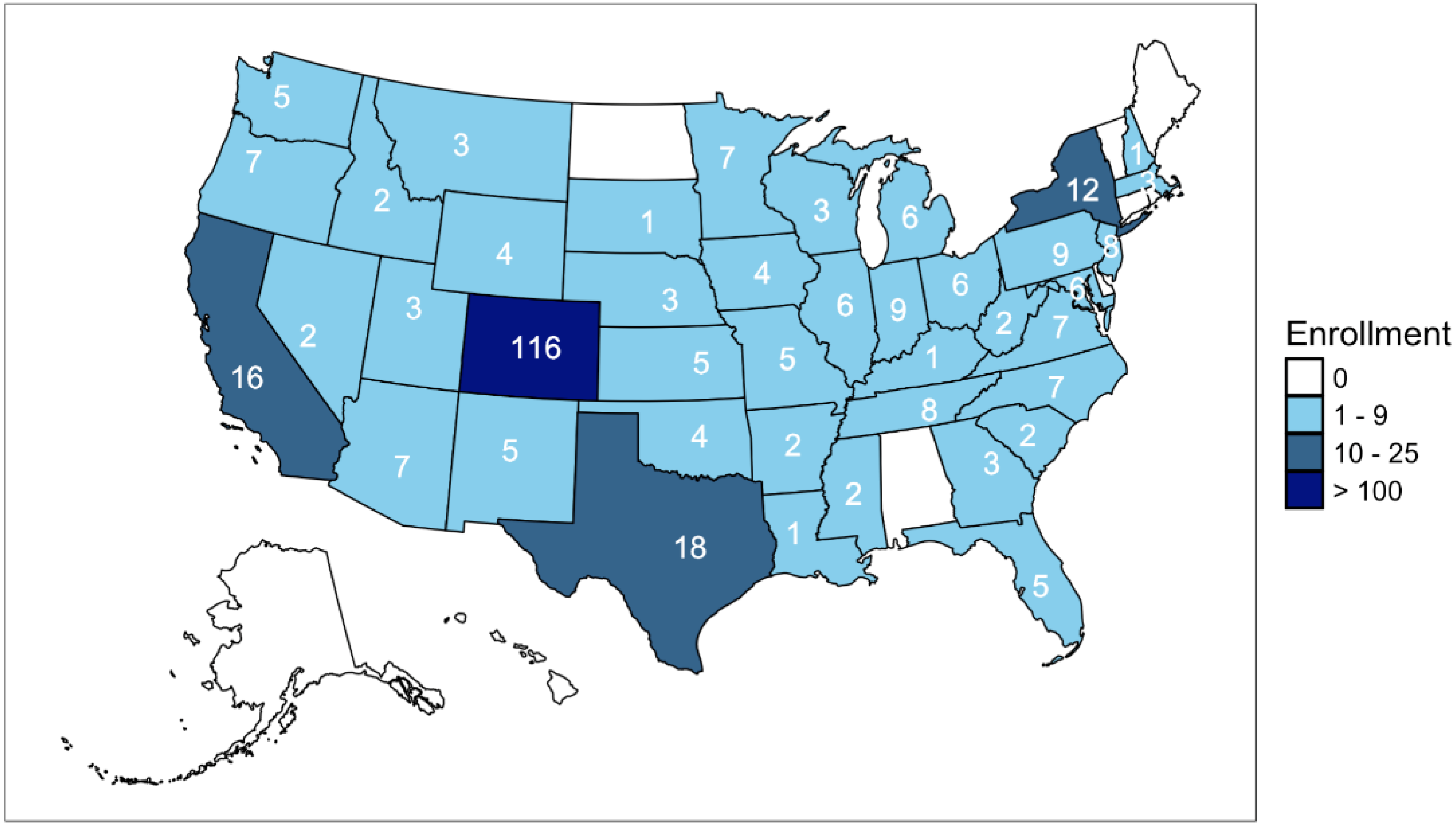
XYY = 34

XXX = 44

XXYY = 74

Other = 26

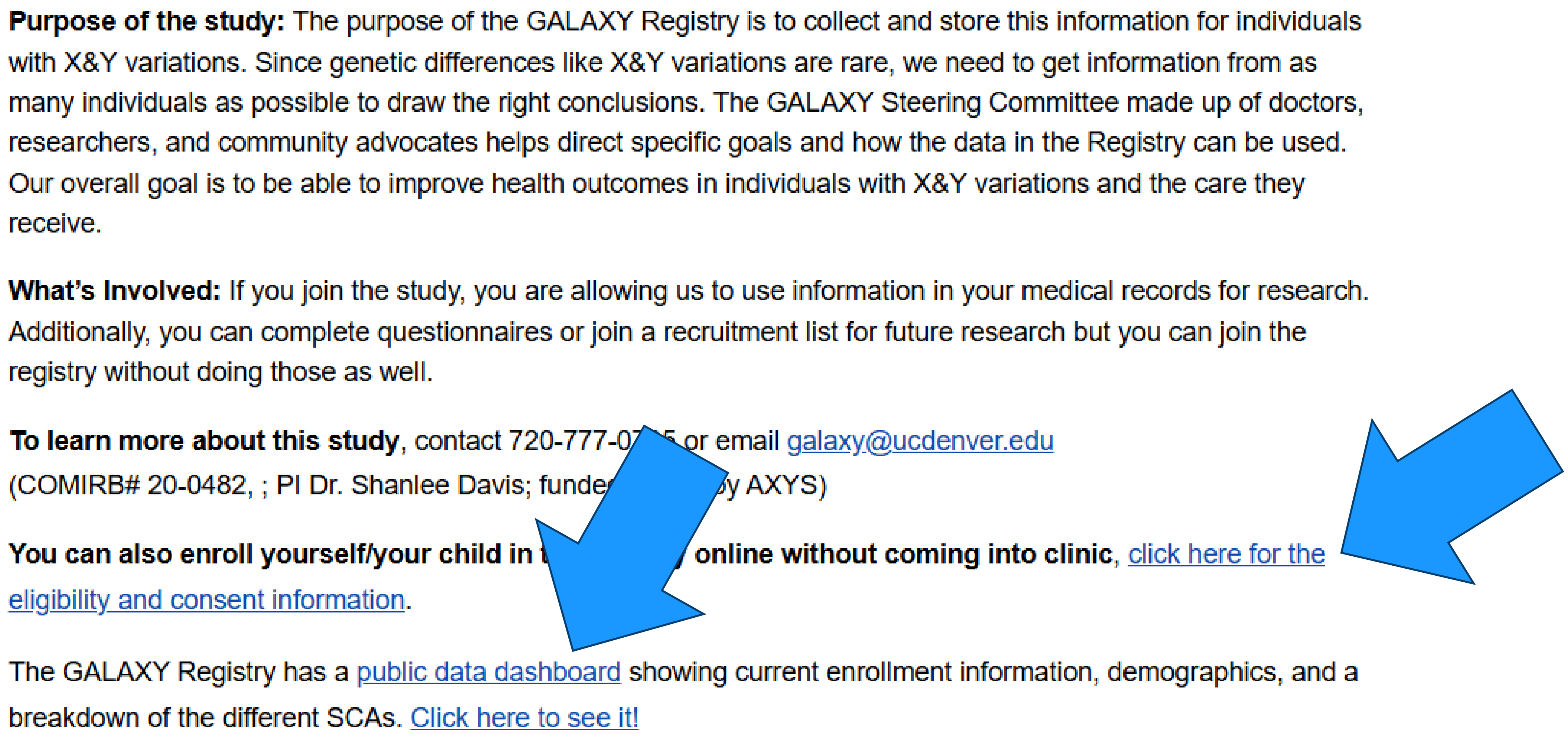
US participants enrolled by state



GALAXY Dashboard

”Live” summary of GALAXY data publicly available at all times

galaxyregistry.org



Purpose of the study: The purpose of the GALAXY Registry is to collect and store this information for individuals with X&Y variations. Since genetic differences like X&Y variations are rare, we need to get information from as many individuals as possible to draw the right conclusions. The GALAXY Steering Committee made up of doctors, researchers, and community advocates helps direct specific goals and how the data in the Registry can be used. Our overall goal is to be able to improve health outcomes in individuals with X&Y variations and the care they receive.

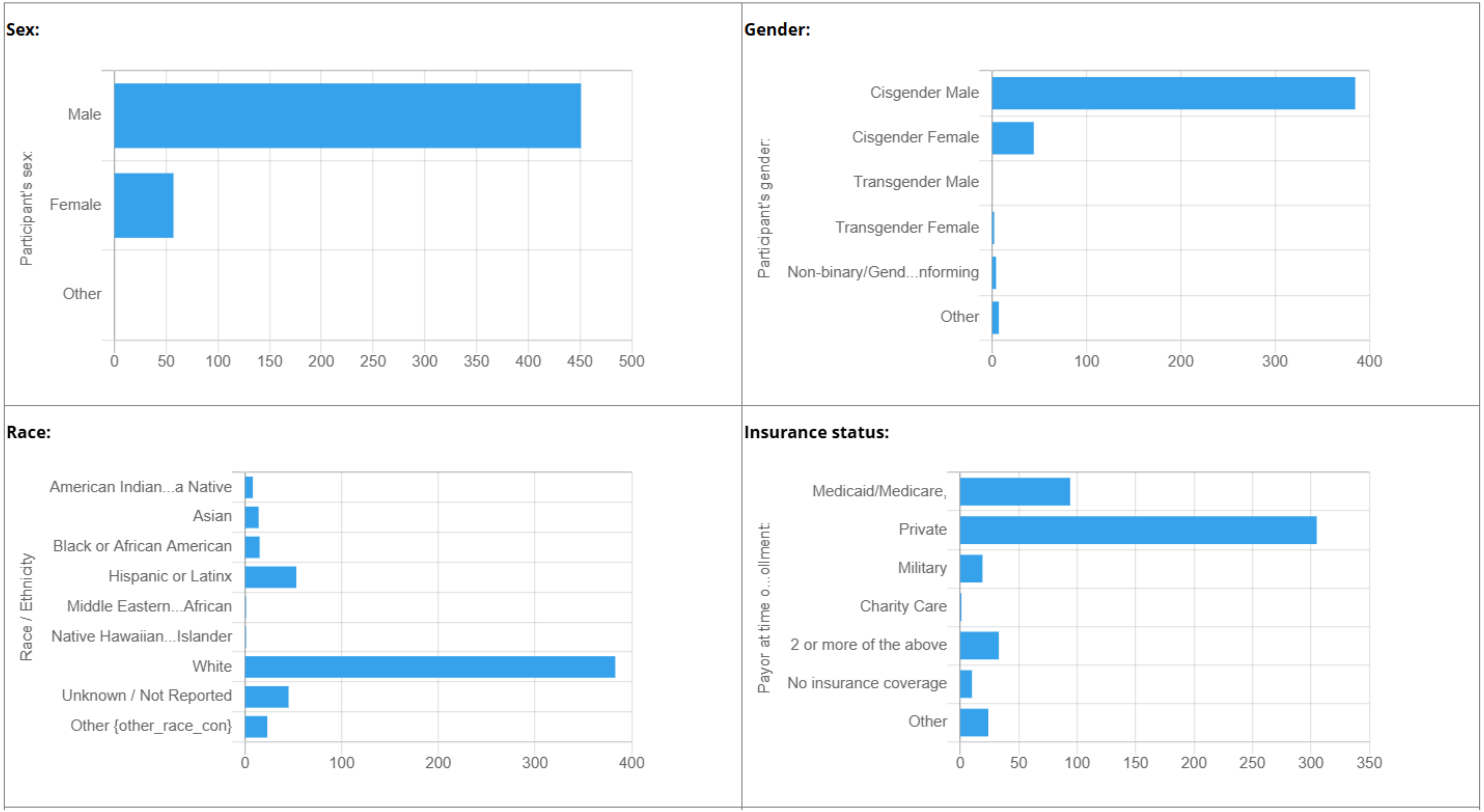
What's Involved: If you join the study, you are allowing us to use information in your medical records for research. Additionally, you can complete questionnaires or join a recruitment list for future research but you can join the registry without doing those as well.

To learn more about this study, contact 720-777-07[illegible]5 or email galaxy@ucdenver.edu
(COMIRB# 20-0482, ; PI Dr. Shanlee Davis; funde[illegible]y AXYS)

You can also enroll yourself/your child in t[illegible] online without coming into clinic, click here for the eligibility and consent information.

The GALAXY Registry has a public data dashboard showing current enrollment information, demographics, and a breakdown of the different SCAs. Click here to see it!

GALAXY Dashboard



SUMMARY OF SPECIFIC PROJECTS

PAPERS

- Research priorities of the X&Y community
- Psychopharmacology for XXYY
- GALAXY methods

ABSTRACTS

- Initial data from the GALAXY Registry
- Prenatal health in sex chromosome aneuploidies

IN PROGRESS

- Updating the phenotype of XXYY

Research Priorities of Individuals and Families With Sex Chromosome Aneuploidies

Goal:

1

determine the health areas of highest research priority to the SCA community

2

explore differences in priorities when stratified by SCA group

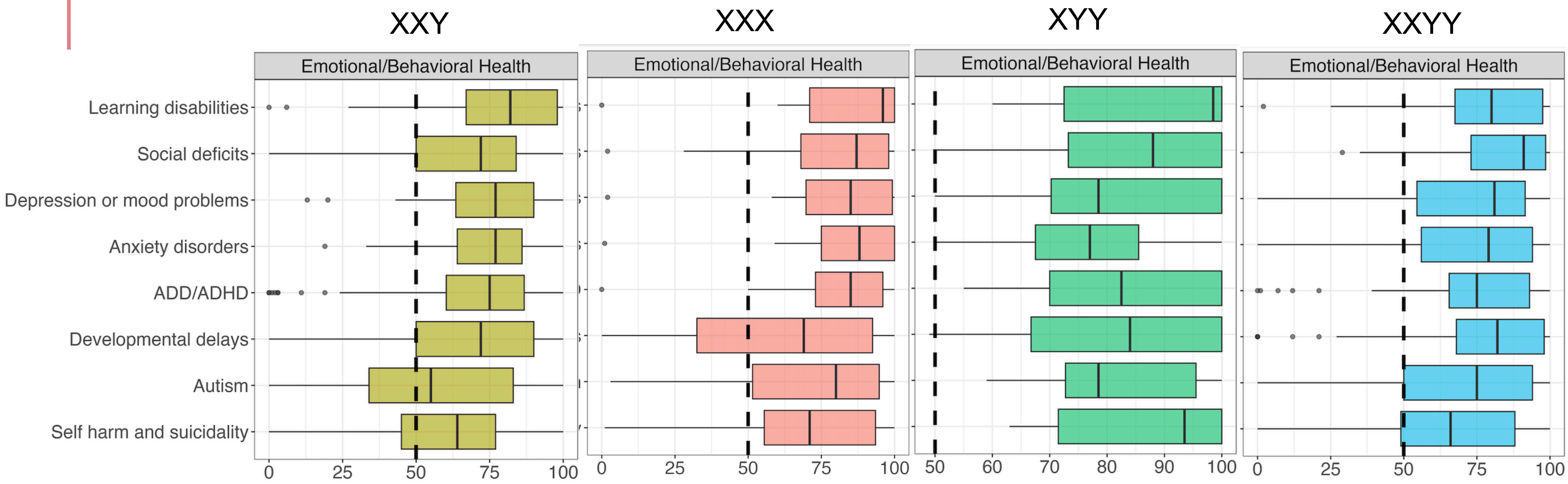
3

Explore differences when compared by respondent (self-advocate versus caregiver)

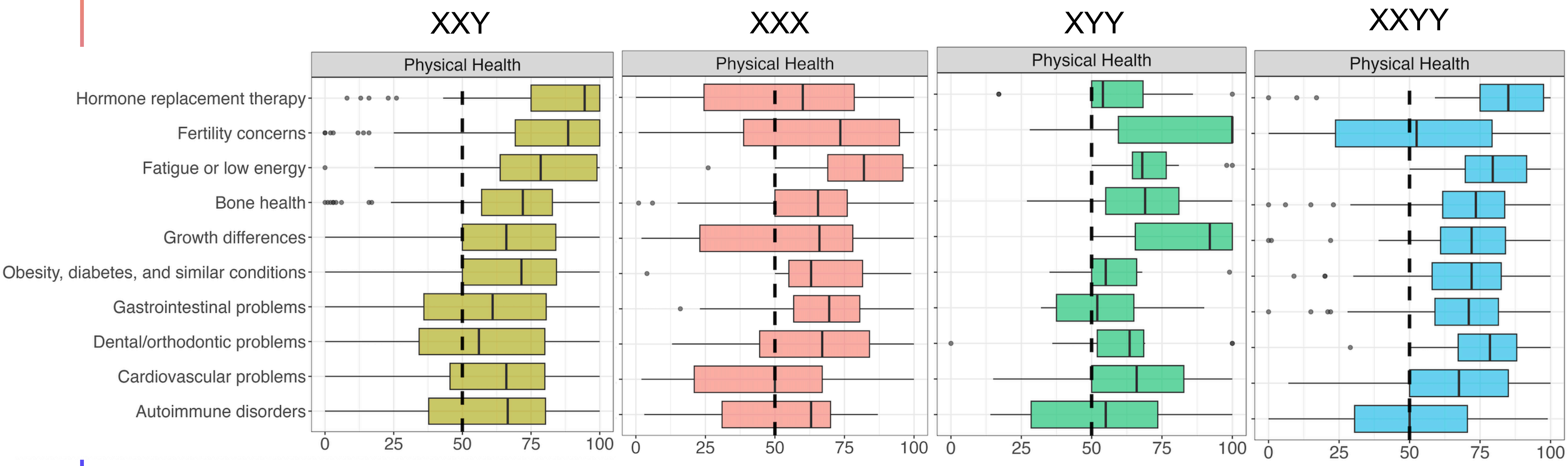
Who answered the survey?

	XXX (*N* = 17)	XXY (*N* = 86)	XYY (*N* = 12)	XXYY (*N* = 35)	All other SCAs (*N* = 9)
Age (mean SD)	14 [5, 22]	10 [1, 20]	3 [2, 9.5]	11.5 [5, 25]	8 [5, 14]
Race/ethnicity (% white non-Hispanic)	12 (70.6%)	76 (88.4%)	8 (66.7%)	29 (82.9%)	9 (100%)
Parent	14 (82.4%)	69 (80.2%)	12 (100%)	35 (100%)	8 (88.9%)
Self-Respondant	3 (17.6%)	17 (19.8%)	0 (0%)	0 (0%)	1 (11.1%)

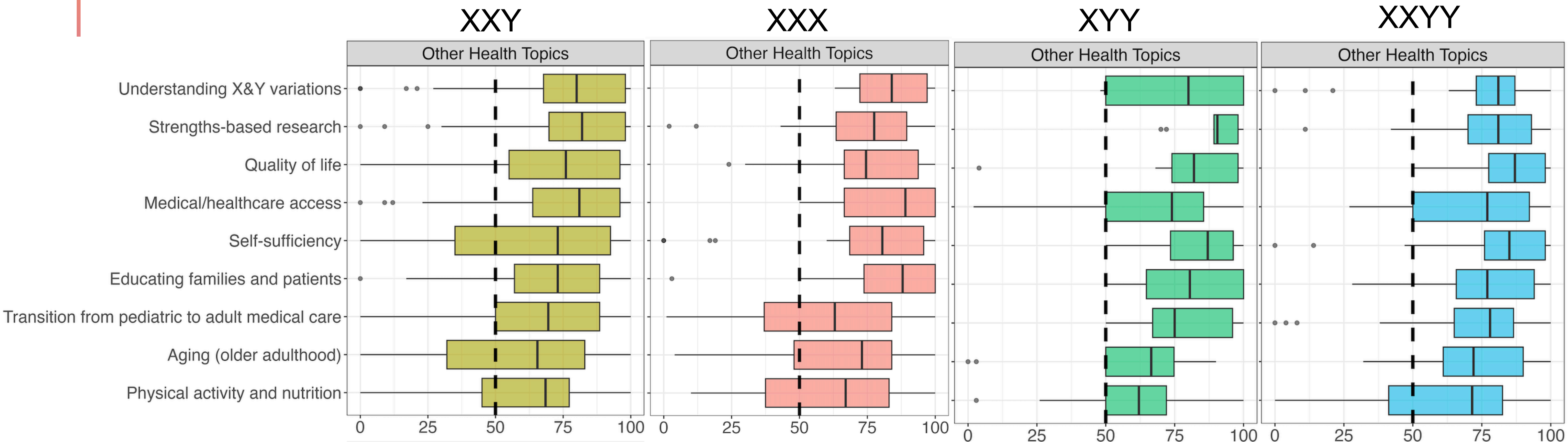
Behavioral Health Priorities



Physical Health Priorities



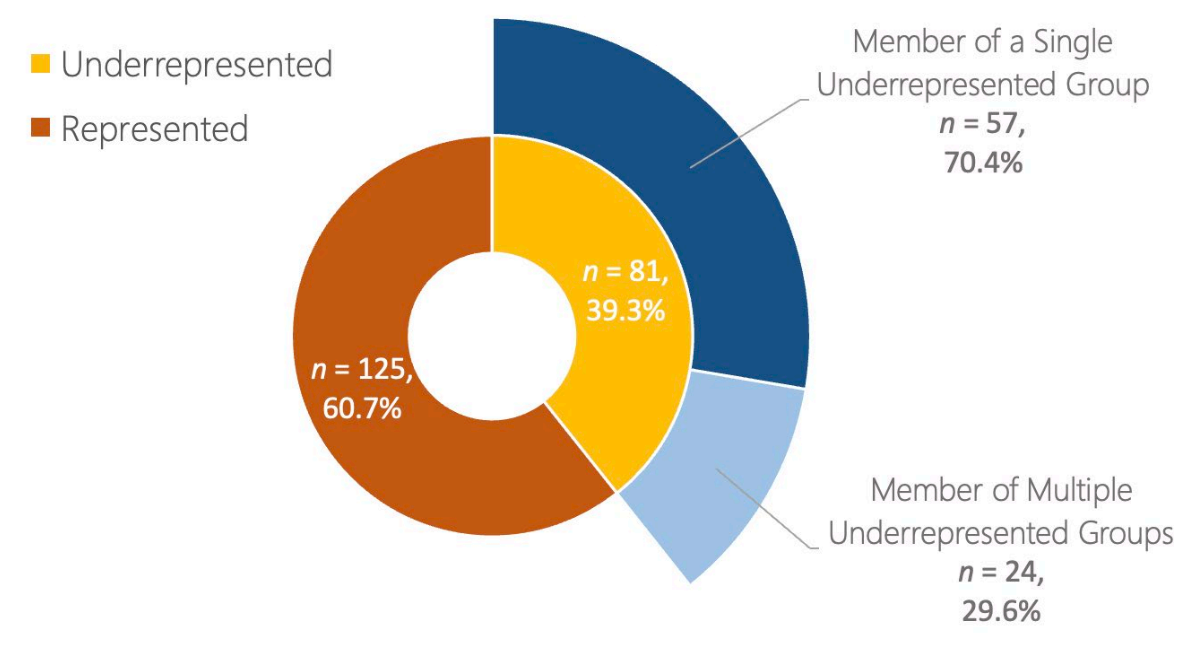
Other Priorities



Generating Advancements in Longitudinal Analysis in X and Y Variations: Rationale, Design, and Methods for the GALAXY Registry

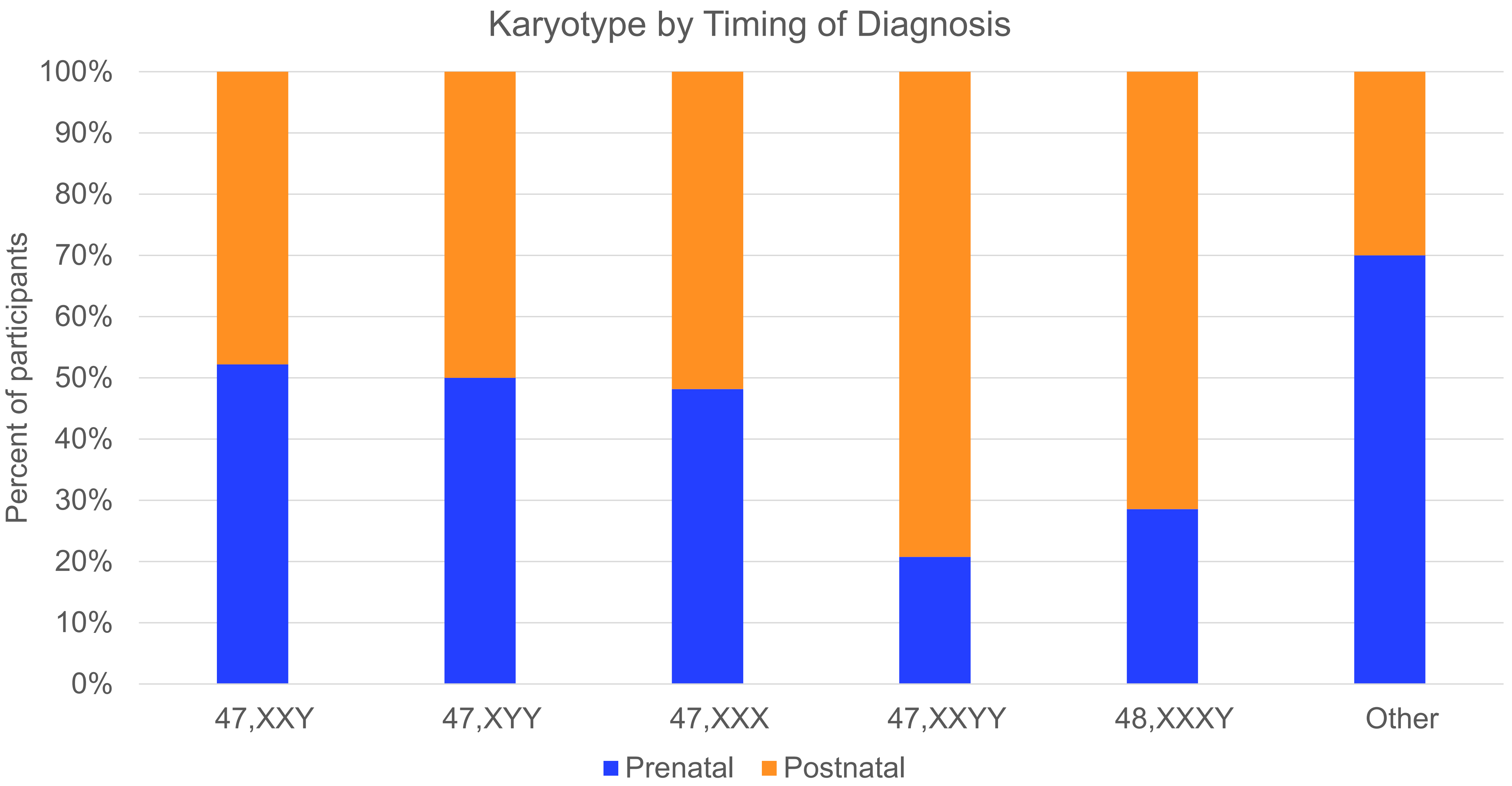
INITIAL DATA FROM THE GALAXY REGISTRY: GENERATING ADVANCEMENTS IN LONGITUDINAL ANALYSIS IN X & Y CHROMOSOME VARIATIONS

- Underrepresented = member of at least 1 underrepresented-in-research group (in categories of race, ethnicity, payor status, gender, and survey language)
- 39.3% belonged to at least one underrepresented-in-research group

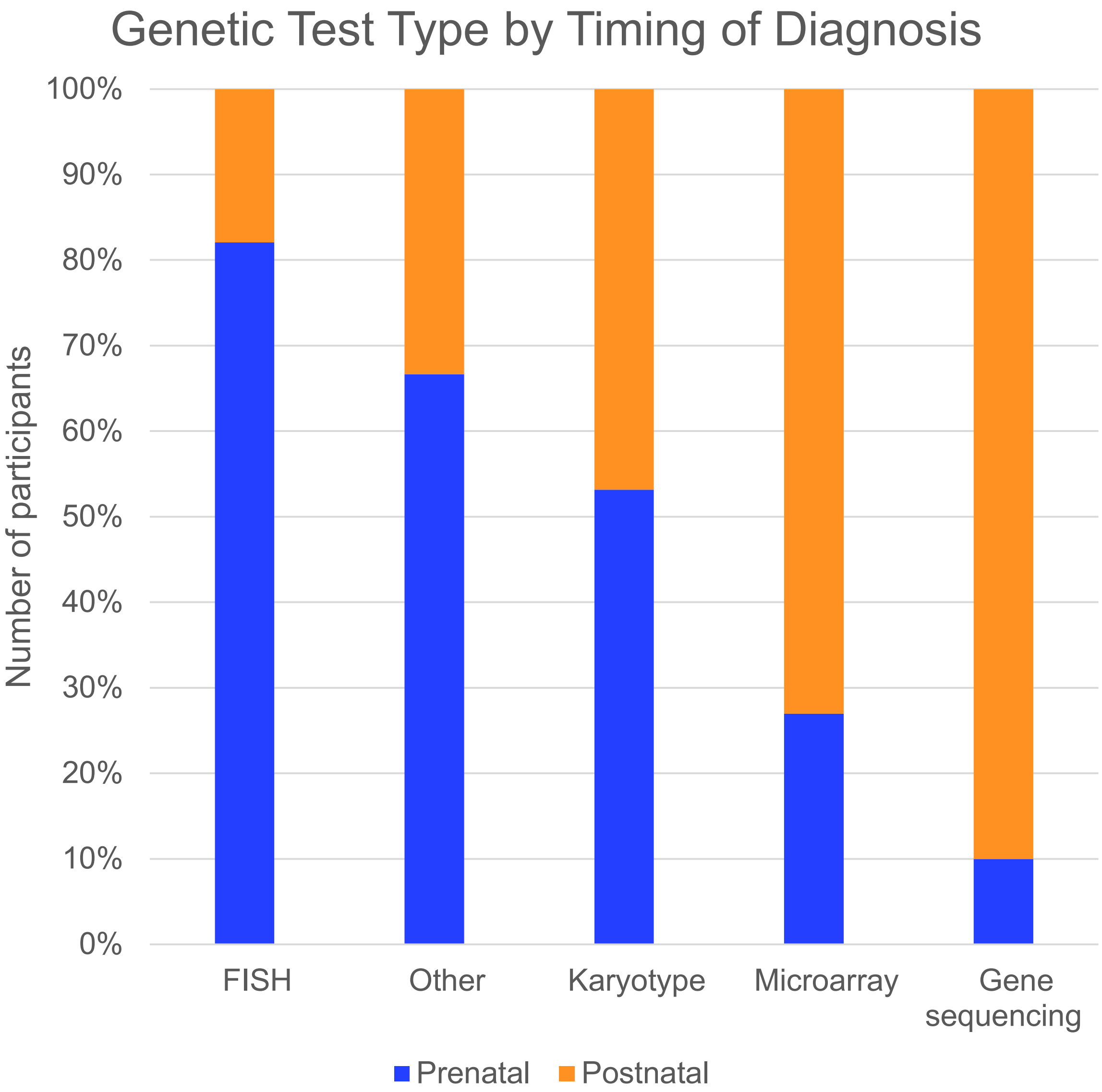


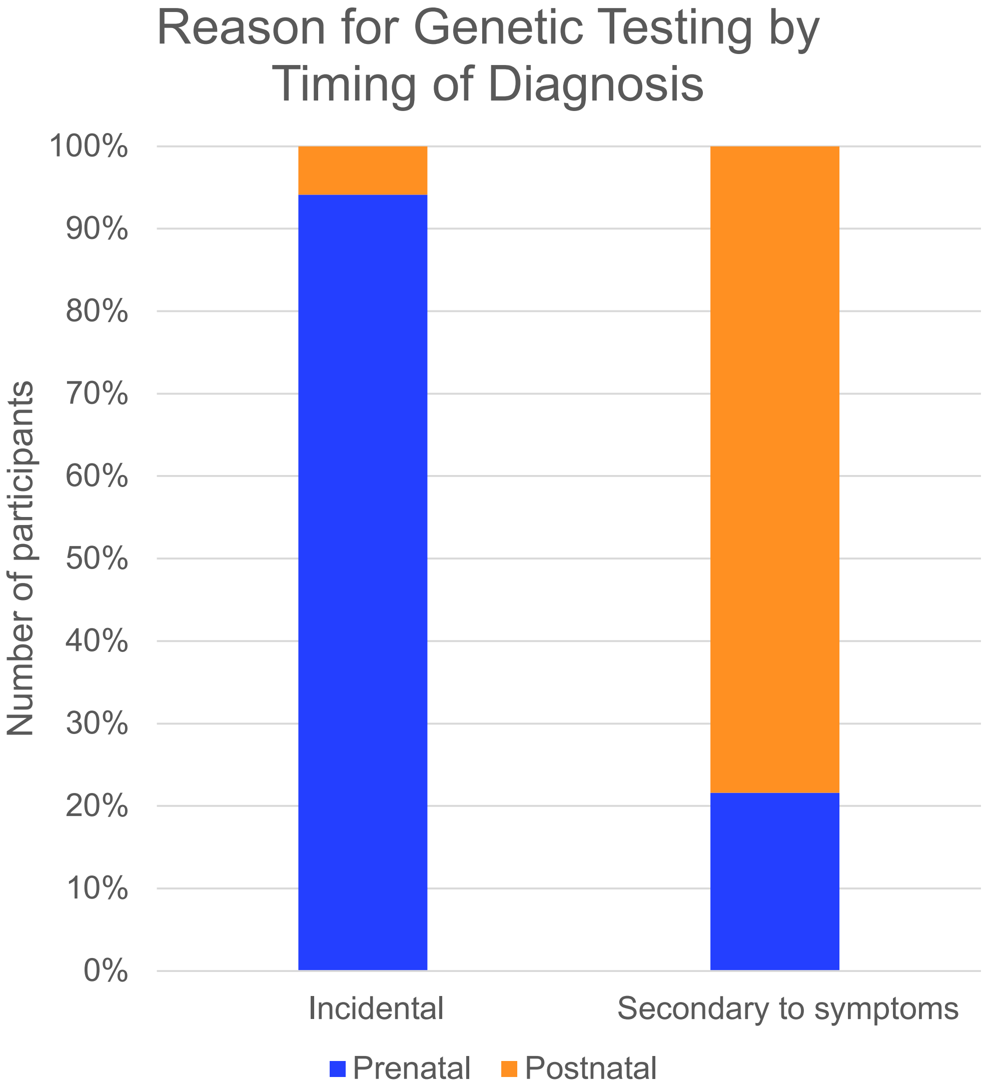
Presented by Kayla Molison at the Western Medical Research Conference 2024

DIAGNOSTIC INFORMATION

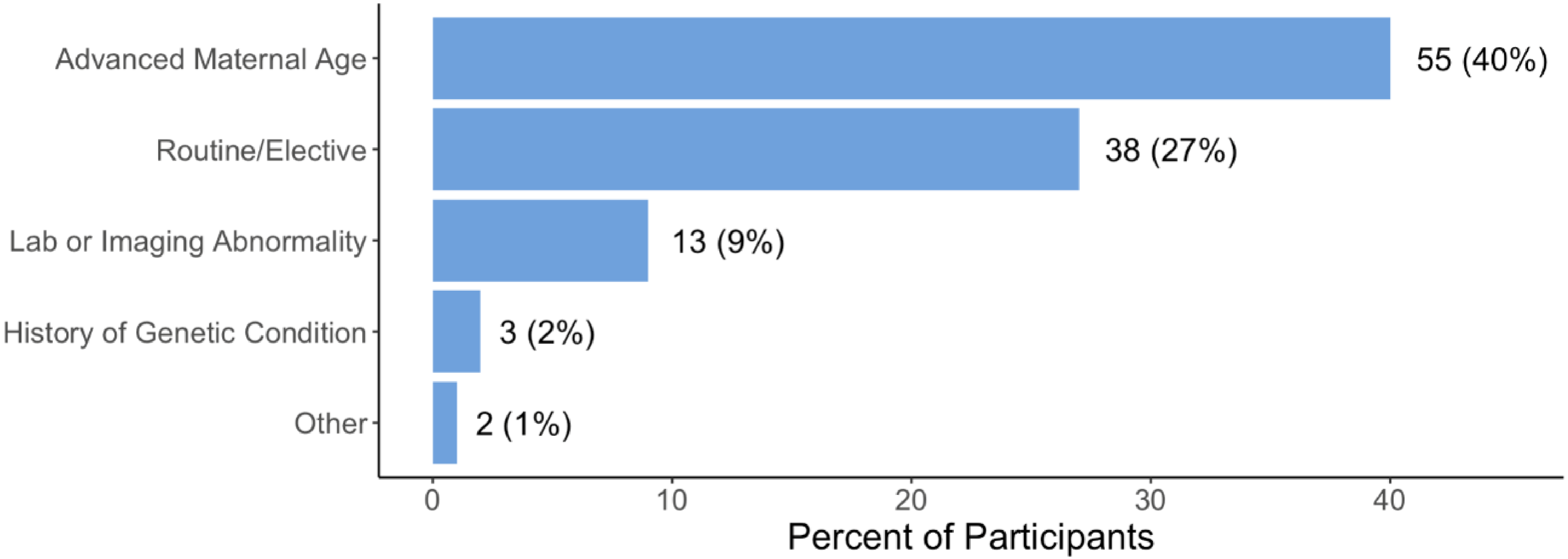


DIAGNOSTIC INFORMATION

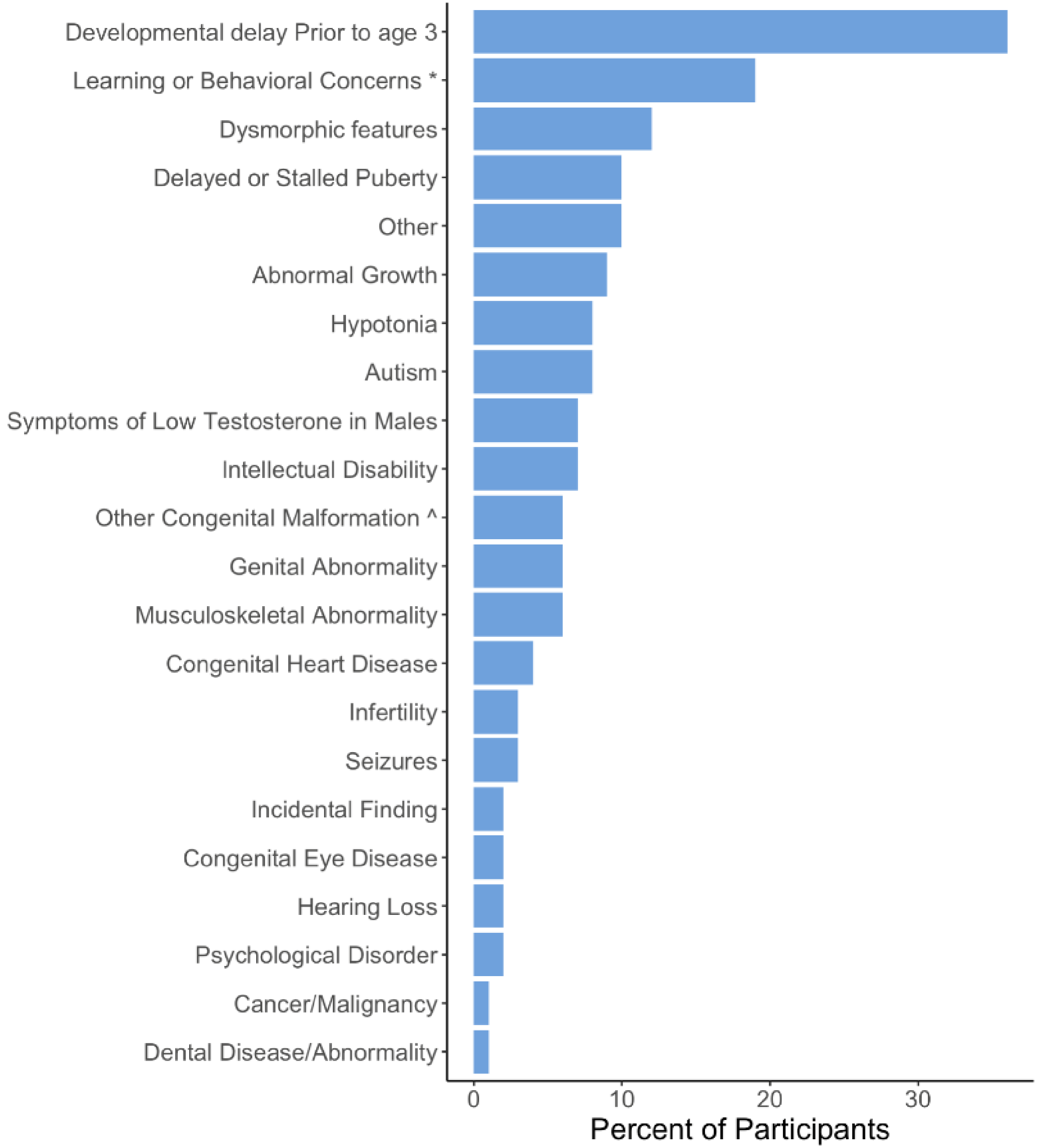




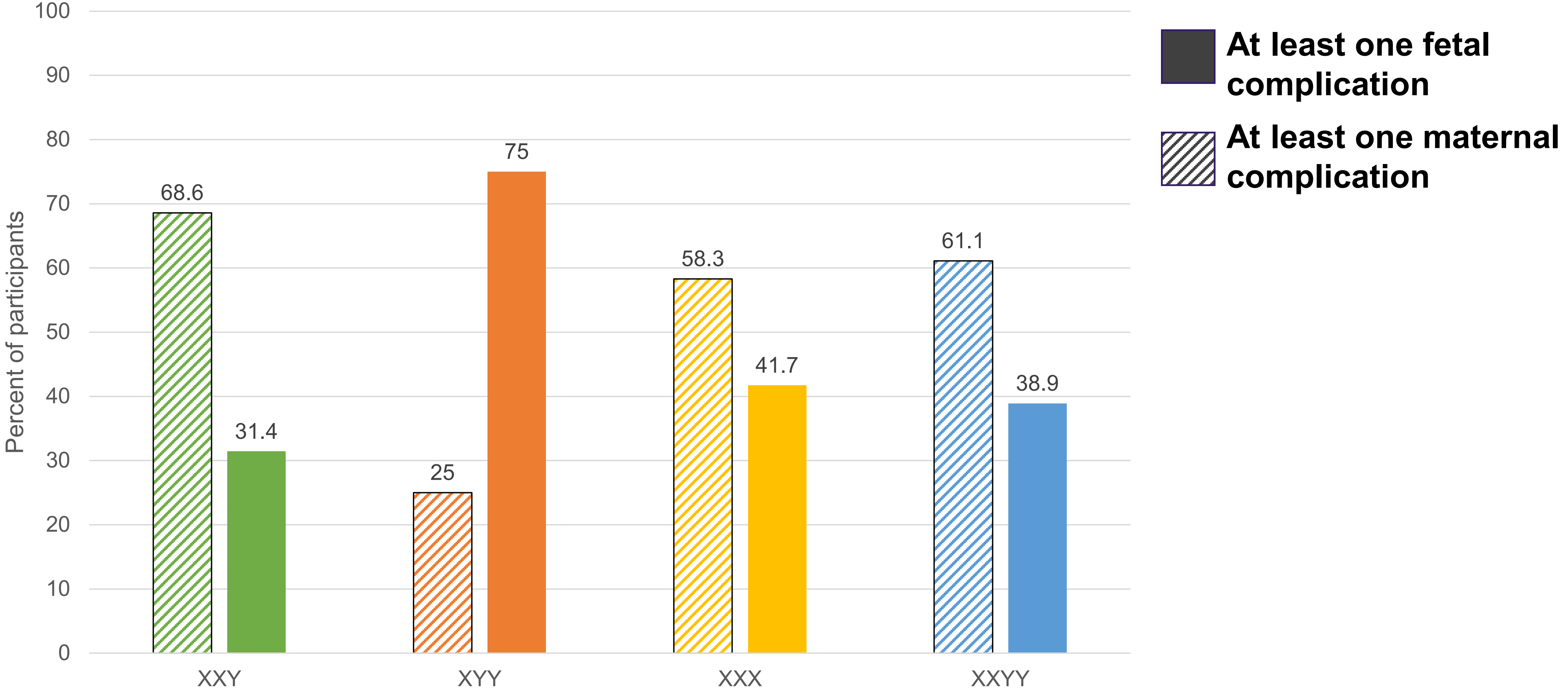
Reasons for Prenatal Diagnoses



Reasons for Postnatal Diagnoses

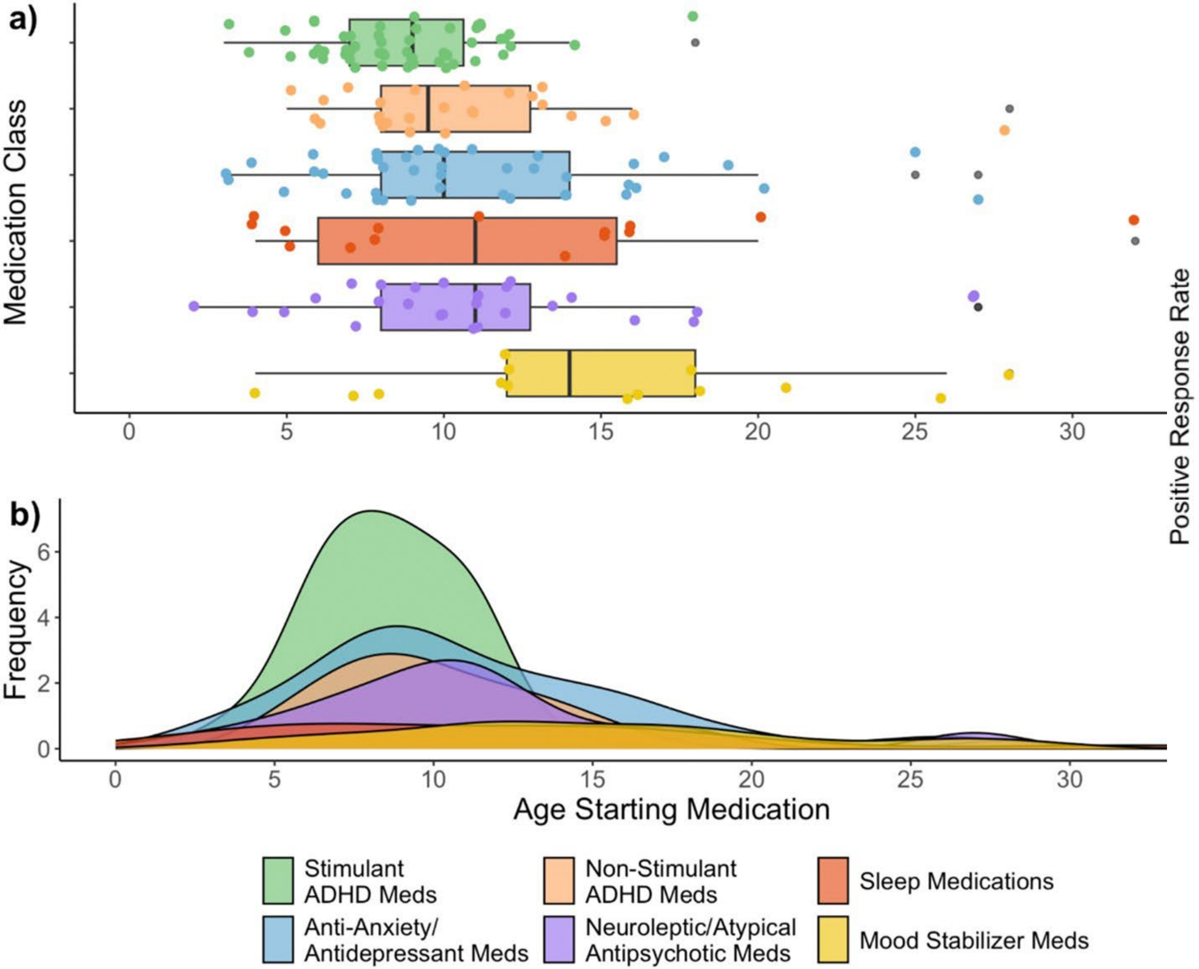


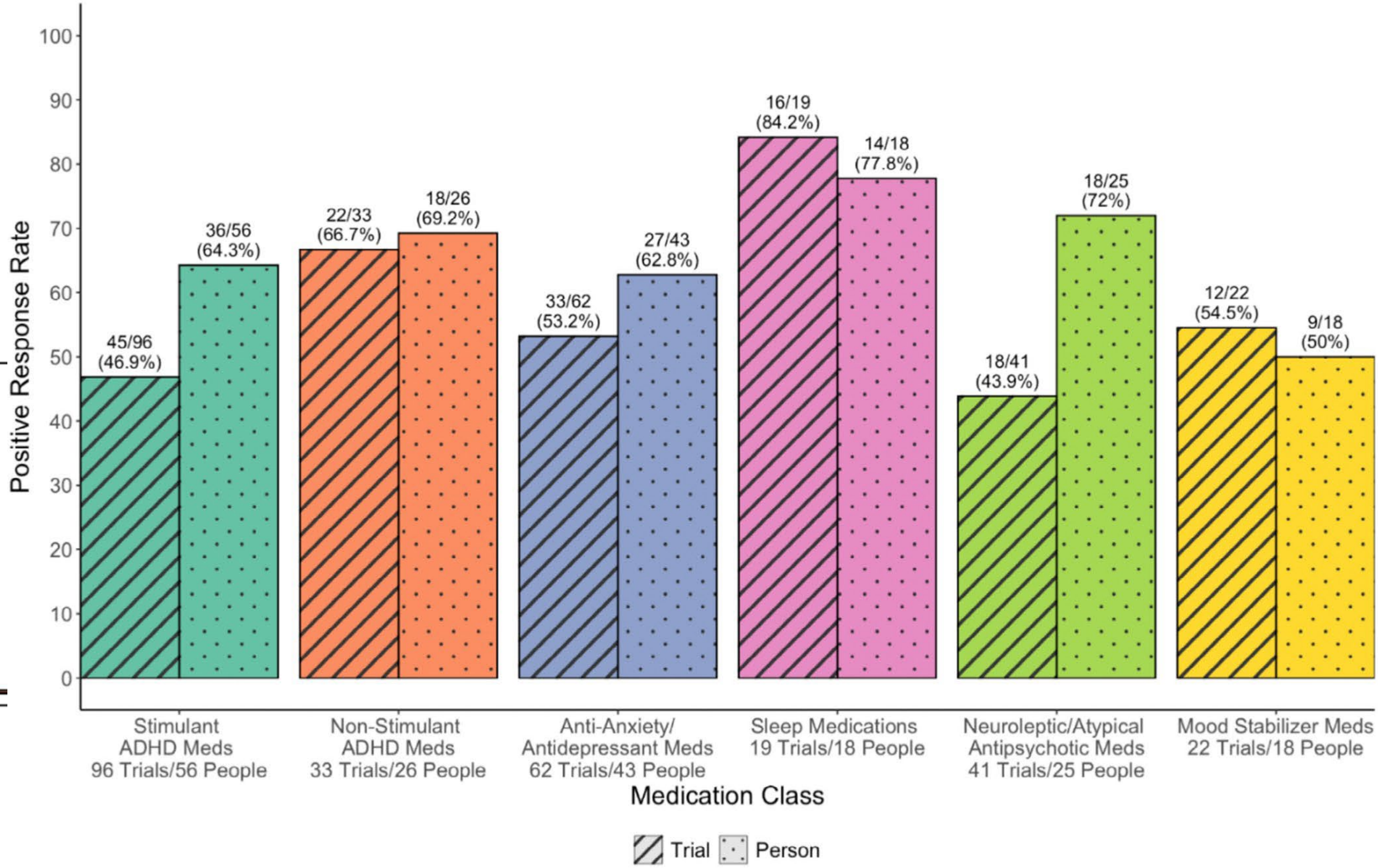
PRENATAL HEALTH IN SEX CHROMOSOME ANUPLOIDIES



Presented by Alyssa Keller at the Western Medical Research Conference 2025

Psychotropic Medication Use in 48,XXYY Syndrome


a)
Medication Class
0
5
10
15
20
25
30
b)
Frequency
0
2
4
6
0
5
10
15
20
25
30
Age Starting Medication
Stimulant ADHD Meds
Non-Stimulant ADHD Meds
Sleep Medications
Anti-Anxiety/ Antidepressant Meds
Neuroleptic/Atypical Antipsychotic Meds
Mood Stabilizer Meds


Positive Response Rate
0
10
20
30
40
50
60
70
80
90
100
45/96 (46.9%)
36/56 (64.3%)
22/33 (66.7%)
18/26 (69.2%)
33/62 (53.2%)
27/43 (62.8%)
16/19 (84.2%)
14/18 (77.8%)
18/41 (43.9%)
18/25 (72%)
12/22 (54.5%)
9/18 (50%)
Stimulant ADHD Meds 96 Trials/56 People
Non-Stimulant ADHD Meds 33 Trials/26 People
Anti-Anxiety/ Antidepressant Meds 62 Trials/43 People
Sleep Medications 19 Trials/18 People
Neuroleptic/Atypical Antipsychotic Meds 41 Trials/25 People
Mood Stabilizer Meds 22 Trials/18 People
Medication Class
Trial
Person

Conclusions

- Medication use is common: 70% currently or having taken medications for a behavior or mental health indication
- ADHD and anxiety/depression were the most common target symptoms
- Stimulant ADHD medications were the most used class, followed by anti-anxiety/antidepressant medications
- Neuroleptics (also called atypical antipsychotics) are often used for treatment of irritability and/or aggressive behavior, and we found that 35.2% of participants had taken neuroleptic medications

WHERE ARE WE GOING FROM HERE?

NASCAR

NETWORK FOR ADVANCING

CHROMOSOME ANEUPLO

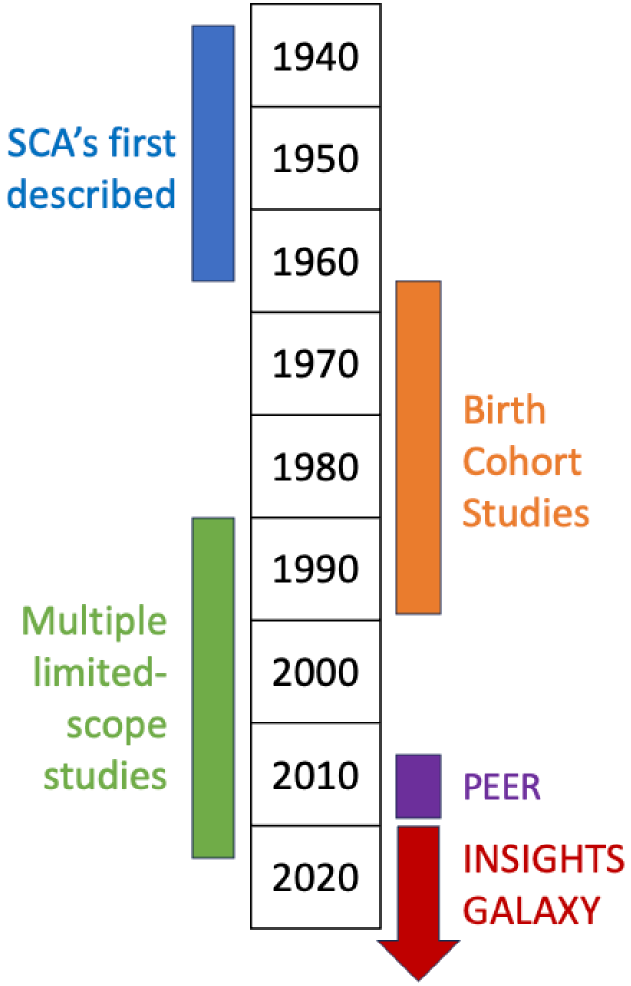
RESEARCH

NASCARR Proposal

- NIH program supporting rare disease consortia
- Application scored very well!
- Project 1: **Longitudinal Registries to Inform the Natural History of Sex Chromosome Aneuploidies**
- 18+ sites across the US

Project 1 Aims

1. Develop a high-yield longitudinal clinical data and biospecimen repository representative of all individuals with SCA utilizing the foundation of the INSIGHTS and GALAXY registries
2. Describe the systemic morbidity associated with each SCA across the lifespan
3. Determine phenotypic traits that are common among all SCAs (or SCA subcategories), as well as those that are unique to individual SCA conditions



SCA's first described

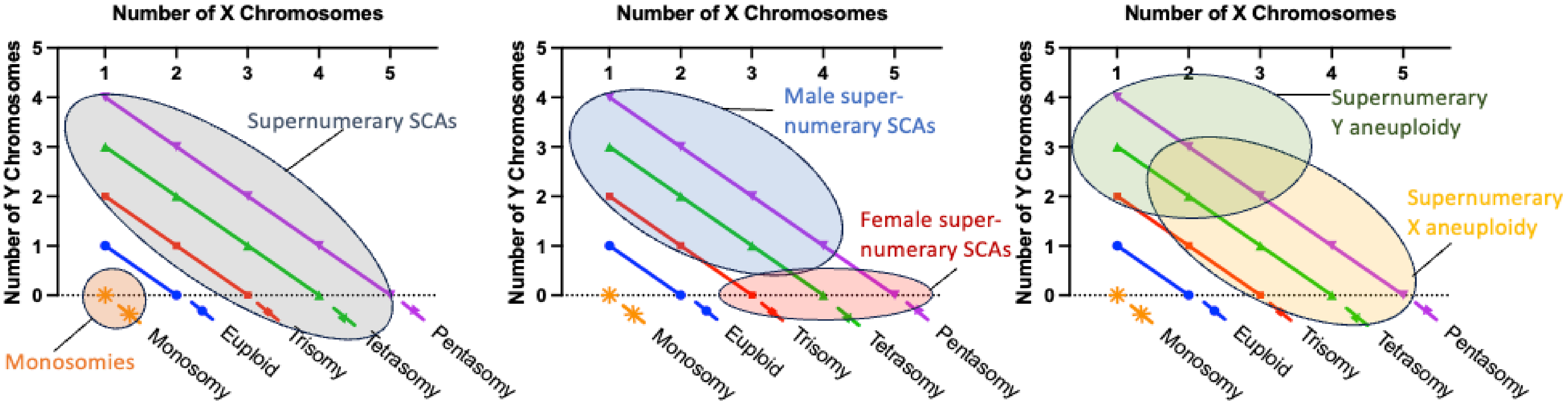
Multiple limited-scope studies

Birth Cohort Studies

PEER

INSIGHTS
GALAXY

More complex analyses



We need >1000+ individuals! HUNDREDS in each karyotype group.

AUDIENCE PARTICIPATION

How can we improve recruitment for our smaller groups to make sure GALAXY is representative of everyone?

WHO WILL ENGAGE NEXT?

Frequently Asked Questions

Why is it important or worth it for me to sign up?

Each person with an X and Y variation is unique! It is important that the Registry represent everyone’s story.

X and Y variations are rare. The more participants we have, the more confident we can be that what we find is true.

Frequently Asked Questions

How do I know my data is private or secure?

1. We follow all federal, state and University rules, laws, and regulations to protect your privacy.
2. GALAXY is overseen by a regulatory board called the *Colorado Multiple Institutional Review Board* that makes sure we are following all the rules and regulations.
3. All data are stored in *Research Electronic Data Capture,* or REDCap. This is a HIPAA-compliant, password protected, online database designed to store secure research information.
4. We use a study ID system where each participant is assigned a number. Most people will just see that number, and not your name.
5. Only individuals approved by the PI with required training have access to REDCap.
6. When we present or publish information from the registry, we group data together to give summary information.
7. Sponsors (including AXYS, XXYY Project, NIH, and any others) do not have access to identifiable data.

Frequently Asked Questions

What is the minimum amount I have to do?

1. Sign the consent form online
2. Upload your genetic test OR tell us where it was done at and give us permission to get it for you

Frequently Asked Questions

Why do you need my genetic test results?

1. We need to make sure you have a genetically confirmed X&Y variation
2. We need to know the exact karyotype (multiple cell lines?)
3. It also contains useful information, such as the date and what symptoms (if any) led to genetic testing

Frequently Asked Questions

What if I don't have the genetic test that shows I have an X&Y variation?

You can sign a Release of Records. This allows the study team to reach out to the clinic where you had the test done at and we can get a copy that way instead.

- Chromosome analysis (karyotype) from CVS, amnio or postnatal (any tissue)
- FISH from any tissue
- Microarray
- Specific panel identifying X&Y aneuploidy (prefer follow up karyotype)
- Whole exome or genome sequencing

If you do not have a copy of your genetic test results, consider talking to your doctor about ordering as this is important for your records, too.

Frequently Asked Questions

What else is involved? Optional procedures:

- ***Give permission to access medical records.*** The basis of GALAXY is longitudinal clinical data (clinic visits, labs, medications, etc). If you have received care at any place other than a GALAXY clinic, we need your permission to request these records.
- ***Join the future studies list.*** We can contact you with studies that you may be interested in. This is usually only a few emails per year. You can then learn more about the study and decide if you want to participate.
- ***Complete surveys.*** Medical records only tell part of the story. Some information is better coming directly from you! Maximum of 1 new survey/month (usually less)

Frequently Asked Questions

How can I be *more* involved?

- ***Biobank.*** Blood sample at a GALAXY clinic location.
- ***Steering Committee and/or special interest committees.*** We need volunteers willing to lead the future of GALAXY!
- ***Research Proposal.*** Template available on the website.
- ***Funding and/or Donations.*** We desperately need financial resources to continue GALAXY. Know of any donors or organizations who may be interested in supporting this work? Are you able to support this work? We accept contributions of all sizes!

Frequently Asked Questions

I can't remember if I already signed up for GALAXY. How do I know?

Our team is excited to help you check if you’ve already joined the registry and/or if anything is missing.

- Stop by at our table during the conference OR
- Send an email to galaxy@ucdenver.edu with your name and birthdate (or your child’s information). This will help us find your records. If you are a parent of an adult, please have your child reach out to us directly unless you are their Legally Authorized Representative.

CONCLUSION

- GALAXY is a community partnership
- Already generating important information for the X&Y variations
- Just getting started with a lot of future opportunities! Join us!

Sign up online!

redcap.link/GALAXY-consent

We are here to help throughout the conference!

eXtraordinarY Kids Team

Nicole Tartaglia, MD
Susan Howell, MS, CGC
Jennifer Janusz, PhD
Rebecca Wilson, PsyD
Talia Thompson, PhD
Samantha Bothwell, MS
Megan Louderman, PsyD
Caitlyn Middleton, PhD
Syd Martin, MS OTR
Jackie Frazier, MS, CCC-SLP
Michele Martinez-Chadrom, MS, CCC-SLP
Kayla Molison, BS
Kayla Nocon, MS
Lidia Grzybacz, BS
Karli Swenson, PhD
Joanna Dreyer, PharmD
Morgan Jolliffe, PsyD
Cierra Gurley, PA-C
Many amazing students

GALAXY Steering Committee

Dr. Alan Rogol
Dr. Anna Dawczyk
Dr. Armin Raznahan
Dr. David Hong
Gail Decker
Ginnie Cover
Dr. Lilian Cohen
Dr. Shanlee Davis
Steve Gerken
Susan Howell, CGC
Dr. Nicole Tartaglia

Children's Hospital Colorado
Here, it's different.

School of Medicine
UNIVERSITY OF COLORADO
ANSCHUTZ MEDICAL CAMPUS

LIVING WITH XXY

XXYY PROJECT

axys

GALAXY Dashboard

Research Priorities Paper

GALAXY Methods Paper Preprint

Psychopharmacology in XXYY

How can we improve recruitment?

Sign up for GALAXY